CASE REPORT

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Diffuse Skin Tightness in Familial Mediterranean Fever: A Case Report and Review of Literature

Elham Behrangi[1], Nasrin Shayanfar[2], Hadi Mohagheghian Yaghoubi[3], Saman Aghabekloo[4], Zahra Azizian[5]✉

[1] Department of Dermatology, Rasoul-Akram Hospital, Iran University of Medical Sciences, Tehran, Iran
[2] Department of Pathology, Rasoul-Akram Hospital, Iran University of Medical Sciences, Tehran, Iran
[3] Department of Immunology, Shahid Sadughi University of Medical Science, Yazd, Iran
[4] Islamic Azad University, Tehran Medical Sciences Branch, Tehran, Iran
[5] Skin and Stem Cell Research Center, Tehran University of Medical Sciences, Tehran, Iran

Abstract

Introduction: Familial Mediterranean Fever (FMF) is an autosomal recessive inherited disorder that has skin presentations like vasculitis and paniculitis. There has not been shown any association between systemic sclerosis and FMF in studies. Hence, we report a case of FMF with diffuse skin tightness.**Case Presentation:** An 18-year-old girl known as a case of FMF for 3 years presents to our department with tight skin since childhood. The stiffness of skin appeared when she was about 7 years old. In physical examination, tight skin with general induration in all surfaces of skin, sclerodactyly and beaked nose microstomia are noticed. The findings of all hematological, biochemical and pathological studies were normal.**Conclusion:** Finally, it may be concluded that FMF should be considered as a differential diagnosis in patients attending with skin tightness and the possible etiology is cytokines. **[GMJ.2015;4(4):169-72]**

Keywords: Familial Mediterranean Fever; Skin Tightness; Sclerodactyly; Scleroderma.

Introduction

Familial Mediterranean Fever (FMF) is an autosomal recessive inherited and inflammatory disorder characterized by periodic episode of fever, peritonitis, pleurisy and arthritis. The most common symptoms consist of abdominal pain, typical chest pain and myalgia [1, 2]. If left untreated, amyloidosis causes renal failure frequently. Cutaneous involvement of FMF occurs in 7% to 10% of patients. Erysipelas-like erythema is the most common and known skin lesion of FMF; however, other clinical involvements of skin such as vasculitis and paniculitis may be seen in patients with FMF. Some diseases are associated with FMF, which include Behcet's disease [3], IBD and lupus erythematosus [4], but there has not been shown any association between systemic sclerosis and FMF in studies. Hence, we report a case of FMF with Diffuse skin tightness.

Case Presentation

An 18-year-old girl known as a case of FMF for 3 years presents to our department with tight skin since childhood. She does not have any family history of FMF. Its disease diagnose was confirmed by genetic analysis. There

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Fax: +98 731 2227091
PO Box 7461686688
Email:info@gmj.ir

✉ **Correspondence to:**
Zahra Azizian, Skin and Stem Cell Research Center,
Tehran University of Medical Sciences, Tehran, Iran
Telephone number: +989122788611
Email Address: azizian_z@yahoo.com

was hemozygotic mutation in MEFV on chromosome 16. The stiffness of skin was progressive. It first appeared when she was about 7 years old. She had a history of autoimmune hepatitis at age of 6, cochlea implantation at age 8 and also she went under surgery due to accessory left side breast. During past 3 years, she had taken Colchicine irregularly and three flares of disease had occurred but there were no cutaneous lesions coincided with flare of disease or the interval between flares. In physical examination, tight skin with general induration in all surfaces of skin, sclerodactyly (Figure1) and beaked nose microstomia (Figure2) are noticed.

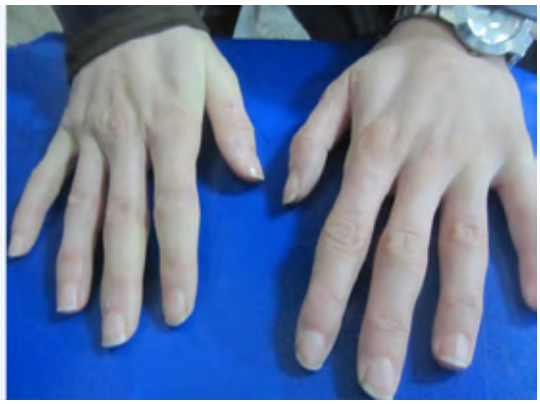

Figure1. Sclerodactyly of all Fingers

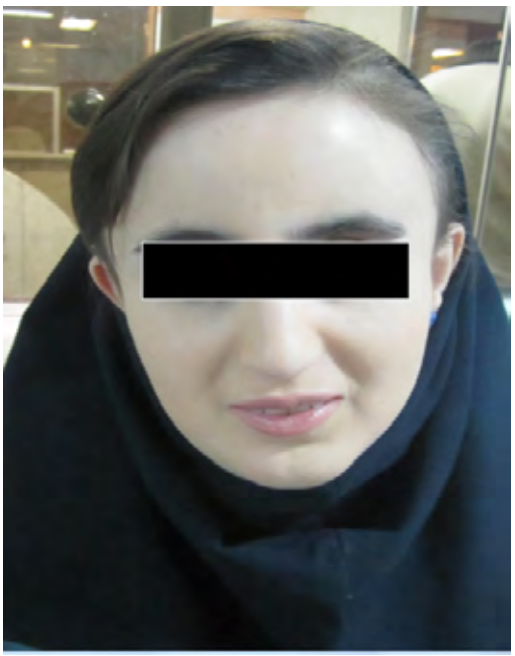

Figure2. Tight face skin can lead to a beaked nose and microstomia.

The clinical features of systemic sclerosis such as Raynaud's phenomenon, digital ulcers and internal organ manifestations all are absent. Special auto antibodies which are positive in systemic sclerosis such as ANA, anti-dsDNA, anti-centromere and anti-scl70 are negative. All differential diagnosis of scleroderma includes skin changes such as gammopathy (by immune electrophoresis), mucin deposition (by biopsy), drug and chemical induced disorders (by history taking) are ruled out. There was no evidence of lung fibrosis in spiral computed tomography (CT) scan and also manometry and barium enema of esophagus was normal. All biochemical and hematological laboratory tests are normal. Histopathology of skin did not show special finding, just little acanthosis and hyperkeratosis. In addition, increased level of elastin and collagen in dermis was present (Figure3). For medical ethics consideration, all figures are presented with eye blinding and with permission of patient.

Discussion

FMF is AR inherited disease caused by mutation in MEFV located on chromosome 16p [5, 6]. One clinical skin lesion of it is erysipelas which is quite common in FMF, although others such as paniculitis and recurrent bullous dermatosis [7] are occasionally described in FMF.

Some mutations in Mediterranean fever gene appear to result in the disease in a vast majority of cases. This gene produces a protein called pyrin or marenostrin. The protein is expressed mostly in neutrophils and may act as an inhibitor of chemotactic factor (C5a) or perhaps of interleukin8 (IL8). Individuals with normal pyrin/marenostrin levels would have the ability to deactivate the chemotactic factors when it is produced in response to an inflammatory process. However, patients with FMF lack such ability resulting in uninhibited activity of the chemotactic factor and episodes of inflammation in the peritoneum, pleura, skin and joints. These also may lead to inflammatory episodes and excess production of amyloids that all would result in skin tightness [8].

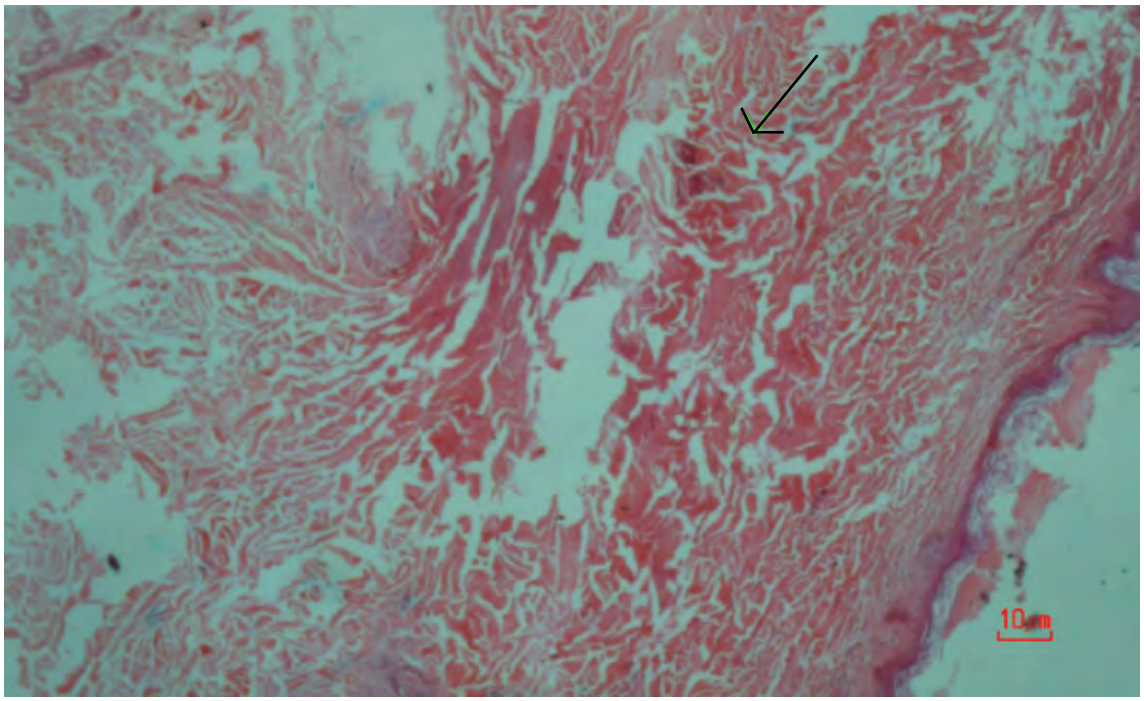

Figure3. Histopathology micrograph of abdomen skin biopsy reveals mild fibrosis (arrow) in superficial dermis with perivascular inflammation (magnification x40, Hematoxylin and eosin)

In a case report by Bafounta *et al*. [9] two unusual skin lesions of FMF which are diffuse; sweet's syndrome like reactions and lichenified erysipelas like are reported. Our report is particular because of atypical scleroderma like skin presentation in a patient with FMF disease. There are some similarities in pathogens of FMF and systemic sclerosis. The cytokines of immune system (IL4 and TNFα) [10-13] which have great roles in collagen synthesis and inducing inflammation are present in both mentioned diseases. This report can lead some authors to hypothesize the effect of mentioned cytokines on scleroderma like skin presentation in FMF disease.

Conclusion

Finally, FMF should be considered as a differential diagnosis in patients attending with skin tightness and the possible etiology is cytokines.

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